PacBio PureTarget Report

This report is generated by software designed to summarize the results of variant, structural variation, and short tandem repeat (STR) genotyping performed using the PacBio PureTarget™ assay. Leveraging high-fidelity long-read sequencing and amplification-free target enrichment, this assay enables accurate characterization of pathogenic repeat expansions and structural variations across a panel of genes implicated in neurodegenerative disease. These results are intended to support heathcare professionals in the diagnostic evaluation and genetic counseling process.

Patient and Report Summary

Patient Name:
Patient Date of Birth:
Sex: **Unknown**
Report Date: **06/13/2025**

Ordering Physician:
Ordering Facility:
Contact/Recipient:

Specimen Site:
Collection Date:
Received Date:

Variant Summary

Gene	Location	Variant	Zygosity	Classification
CYP21A2	Exon 10	NP_000491.4:p.Pro454Ser	Heterozygous	Likely Benign

Tandem Repeat Summary

Locus	Region	Motif	Read Depth	Genotypes		Disease
FRAXE_AFF2	5' UTR	GCC	202 / 201	44	44	⚠ Fragile X syndrome, FRAXE type
FRDA_FXN	Intronic	GAA	319 / 312	18	19	✓ Friedreich ataxia
FXS_FMR1	5' UTR	CGG	200 / 199	28	28	✓ Fragile X syndrome

Symbol Legend

Classification	Symbol
Pathogenic	⊘
Intermediate	⚠
Benign	✓

Primary Findings

AFF2 (INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED)

Tandem Repeat Details

Locus FRAXE_AFF2

Motif	GCC
Genomic Position	X:148500637
Gene & Transcript	AFF2 NM_002025.4
Classification	Intermediate
Location	5' UTR
Read Depth	202 / 201
Genotypes	44 44

Mode of Inheritance: X-Linked Recessive

Prevalence: 1-4/100,000 males ; 1/50-100,000 males, more than 50 families (PMID: 11246464). Found in populations around the globe, including in the UK, US, Canada, Taiwan, Germany, Greece, Cyprus, Spain, and Finland (PMID: 11246464).

Details: Allele ranges (benign:4-39; pathogenic: >200) inferred from The Human Gene Mutation Database. Intermediate alleles correspond to a premutation (PMID: 23914978). Non-canonical motifs include: CGG/CCT/GTG/CAG/CTG3 (PMID: 35245110; PMID: 34111553).

Age at Onset: Typical: 2-10 (PMID: 11246464). Range: 1-10; developmental delays without physical features can make onset difficult to detect until schooling.

CYP21A2 (Congenital Adrenal Hyperplasia, 21-hydroxylase-deficient)

The most common form of congenital adrenal hyperplasia (CAH), characterized by simple virilizing or salt wasting forms that can manifest with genital ambiguity in females and with adrenal insufficiency (in both sexes), and that presents with dehydration, hypoglycemia in the neonatal period (that can be lethal if untreated), and hyperandrogenia.

Variant Details

Variant	NP_000491.4:p.Pro454Ser
Classification	Likely Benign
Location	Exon 10
Allele State	Heterozygous
gnomAD v4 EuropeanNonFinnish Allele Frequency	7005/856458 (0.82%)
NGS Reads Supporting Change	50.40% (249 of 494)

Criteria: BS1, PP2, PS1, BP4

Interpretation: Allele frequency for Annotate-gnomAD Joint Variant Frequencies 4.1, BROAD is above gene threshold. Missense variant in gene with disease commonly caused by missense variants. The variant causes the same pathogenic mutation as a pathogenic variant in the same amino acid for the following sources. Gerp++ predicts tolerated at this location,PhyloP predicts tolerated at this location,Sift predicts this variant is.

Tandem Repeat Classification Thresholds

Pathogenicity classifications for short tandem repeats are based on gene-specific motif count thresholds obtained from the STRchive database, a curated resource of STRs associated with human disease. The ranges below define Benign, Intermediate, and Pathogenic classifications for the reported genes.

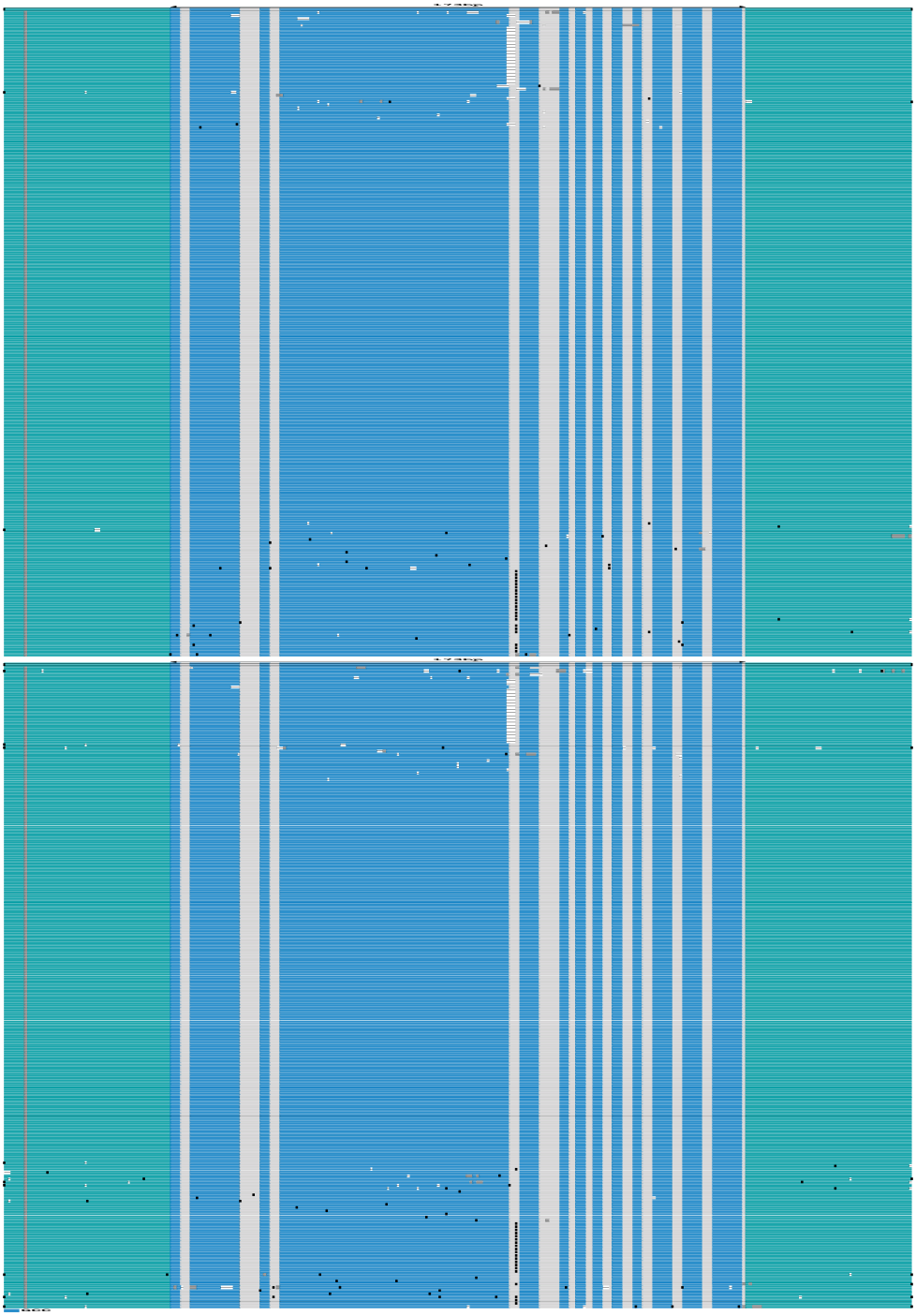
Gene	Disease	Motif	Benign Range	Intermediate Range	Pathogenic Range
AFF2	Fragile X syndrome, FRAXE type	GCC	4 - 39	40 - 200	201 - 2000
FMR1	Fragile X syndrome	CGG	5 - 44	45 - 200	201 - 2000
FXN	Friedreich ataxia	GAA	5 - 33	34 - 55	56 - 1700

Incidental Tandem Repeats

Locus	Region	Motif	Read Depth	Genotypes	
FXN	Intronic	A	319 / 312	16	16
FMR1	5' UTR	AGG	200 / 199	2	2

Supporting Visualizations

FRAXE_AFF2 NM_002025.4 GCC: 44 44



References

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